



estonian genome project



**CARTaGENE
Project**



CIGMR

Public Population Project in Genomics (P³G)

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Overview

The *Public Population Project in Genomics* (P³G) is a not-for-profit and non commercial organization building an international consortium to foster collaboration between researchers in the field of population genomics.

Our Vision:

Foster international harmonization for public projects in population genomics.

Our Goals:

- Understand the interaction between genes, environment, lifestyle and disease
- Create the engine for the transfer of this knowledge to health care systems

Our Approach:

International collaboration so as to ensure public access to population genomics data according to prevailing ethical and legal norms.



Overall Objectives of P³G

Objectives

Provide necessary coordination and harmonization for data collection, production, and storage, to facilitate international collaboration, advance science and maximize public health benefits

Develop common understanding of the socio-ethical and legal issues

Build and maintain a knowledge community for information sharing through a database infrastructure

Foster a deeper understanding of the relative contribution of genetic and non-genetic determinants to health and disease

Transfer this knowledge to the international community so as to optimize benefits for public health care worldwide



Deliverables

Common nomenclature and communication tools required for seamless data sharing

Ethical and legal framework for population-based projects

Public data in a P3G knowledgebase leading to exchange of scientific information and new scientific discoveries in population health research

Discovering linkages between genetics and health

Global access to common data including by developing countries



Partner Projects

GenomEUtwin

Study 800,000 twin pairs from a collaborative European pool of registries through a combination of genetic, epidemiology and phenotype data for common diseases

Estonian Genome Project

Collect data from up to one million participants into a database, including health status, genomic DNA, plasma and genealogical data

CARTaGENE

Obtain personal, medical and sociological data and biological samples from 50,000 random participants for the study of normal genetic variation

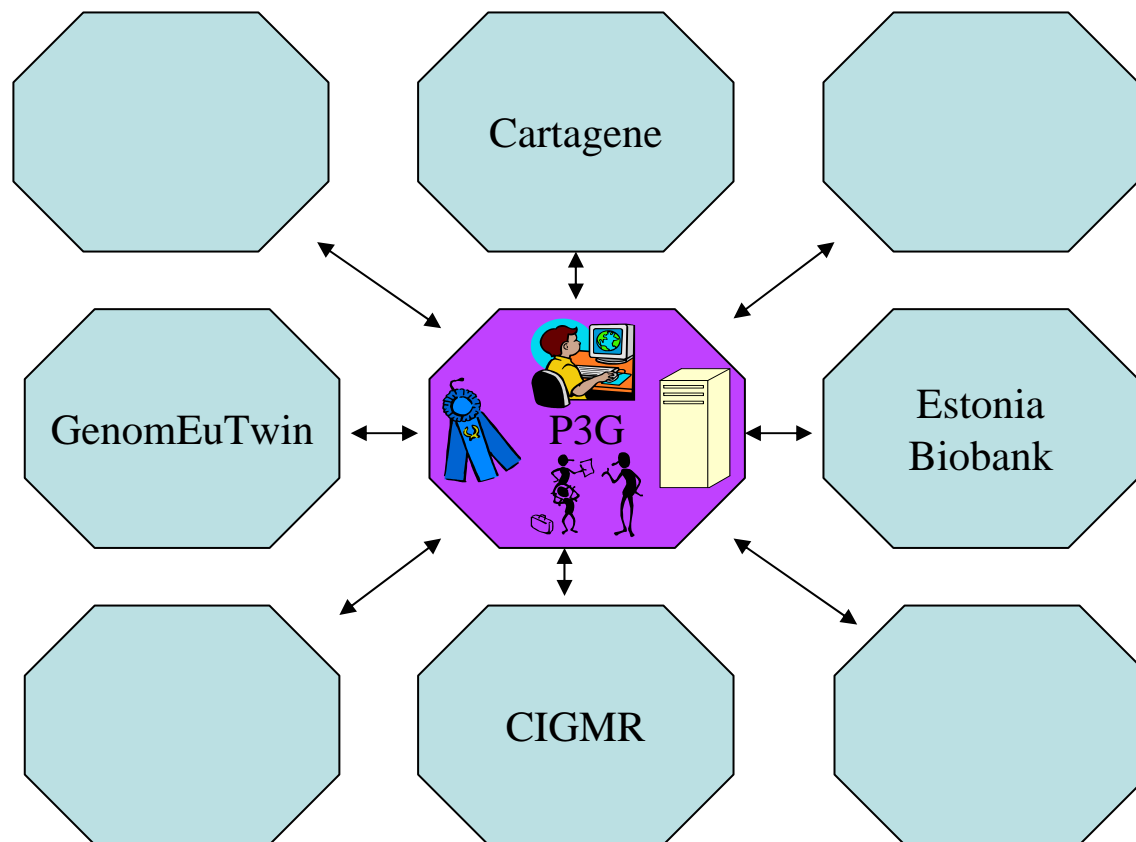
CIGMR

Houses several disease specific population and family based cohorts, and has longitudinal data on over 20,000 individuals



Consortium Model

An international resource for the coordination and exchange of ideas and data that will be generated by the various population biobanks





Areas of Mutual Interest

- **Harmonization** of biological, medical, demographic and social data collected from participants so as to use this information synergistically **in developing effective health care solutions**
- Coordination of possible **access between population databanks** while protecting confidentiality subject to legal constraints, ethical review and governance
- Development of **security measures** for the protection of genetic data and banks in accordance with the highest international standards
- Sharing of approaches to **ethics, public engagement, governance and intellectual property issues**
- **Exchange of experts and young researchers** in many human and social scientific disciplines in addition to biology, public health and genetics
- Determining common biological phenotypes as well as **agreement on methods and quality control**
- Developing **common nomenclature** for data obtained in population genomics projects
- Coordinated development of **bioinformatics** for compatible data mining and for clarity on ownership/copyright issues
- **Support for the transfer of knowledge and technology** to other developed or developing countries
- **Comparative evaluation and validation of research results** and/or hypotheses on health and disease
- **International leadership** in the ethics of genomic and genetic research involving populations



Helsinki/Tallinn Workshops

Goal: Establishing the Work Plan for 2004-2006

I. Socio-Demographic Core

Claude Laberge
Dorret Boomsma

Canada
Netherlands

II. Phenotype Core

Francois Cambien

France

III. Storage, Genotyping and Logistics Core

Leena Peltonen
Andres Metspalu

Finland
Estonia

IV. Public Engagement, Ethics and Governance Core

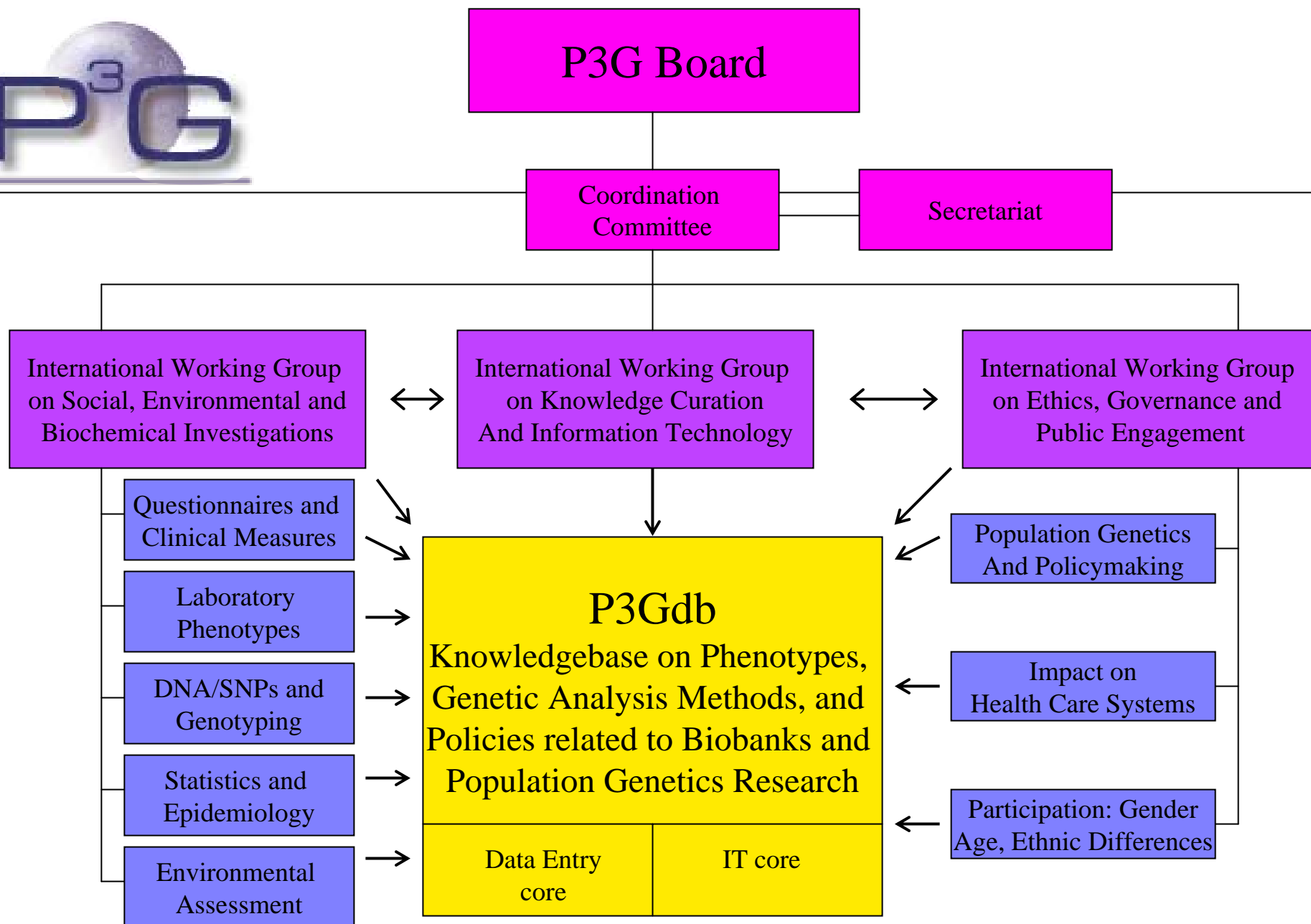
Anne Cambon-Thomsen
Jennifer Harris
Beatrice Godard
Ruth Chadwick

France
Norway
Canada
United Kingdom



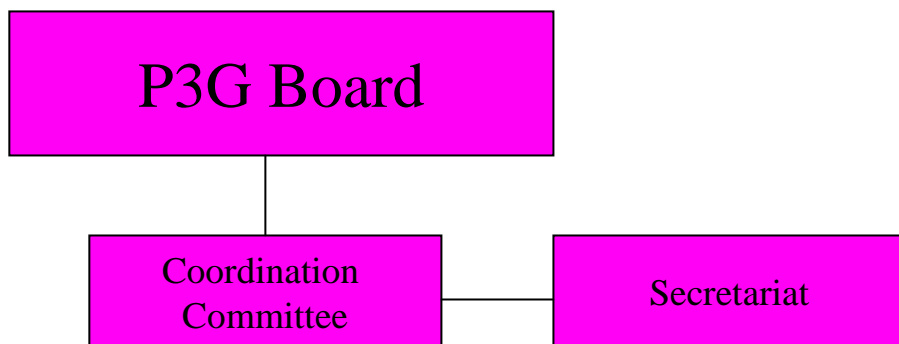
Major Outcomes of the Helsinki/Tallinn/ASHG

- Conclusion that the largest need at present is in better refining phenotypes as opposed to genotypes
- Recommendation to invest early in the creation of a knowledgebase that will track large-scale population genetic studies (including biobanks) - and record their characteristics in order to benefit from existing knowledge and compare approaches (biomedical, ethical, etc)
- Recommendation to merge advisory committees (for easier coordination)
- Definition of core activities for 2004-2006





P3G Governance

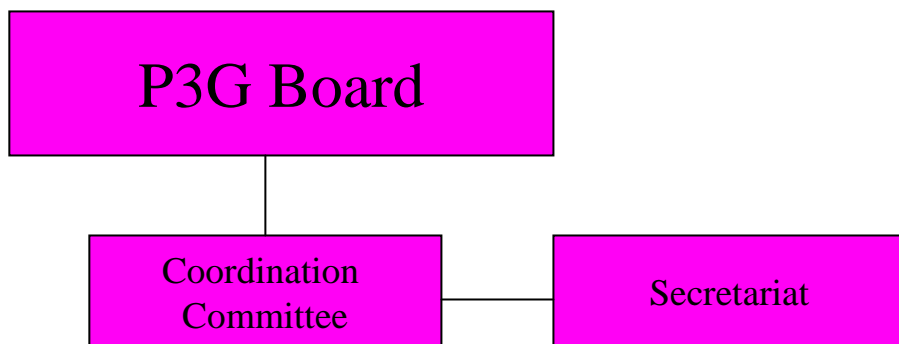


Role of Board :

- ❖ Assure that P3G activities meet the stated goals of harmonization, communication, and public access of P3G deliverables
- ❖ Communication among partners (and funding agencies)
- ❖ Accountability of P3G fund



P3G Governance

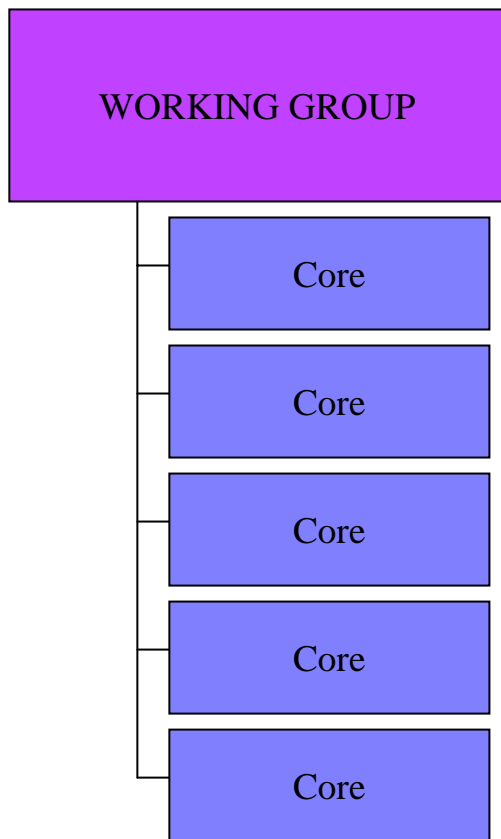


Coordination Committee :

- ❖ Composition includes leaders of Working Group and Cores and BioBank representatives
- ❖ Key role in managing P3G activities, assuring coordination, addressing new issues, etc.
- ❖ Organization of P3G symposia, workshops, exchanges
- ❖ Creating a P3G training program
- ❖ Supported by a small administrative staff



P3G Working Groups and Cores



Working Group

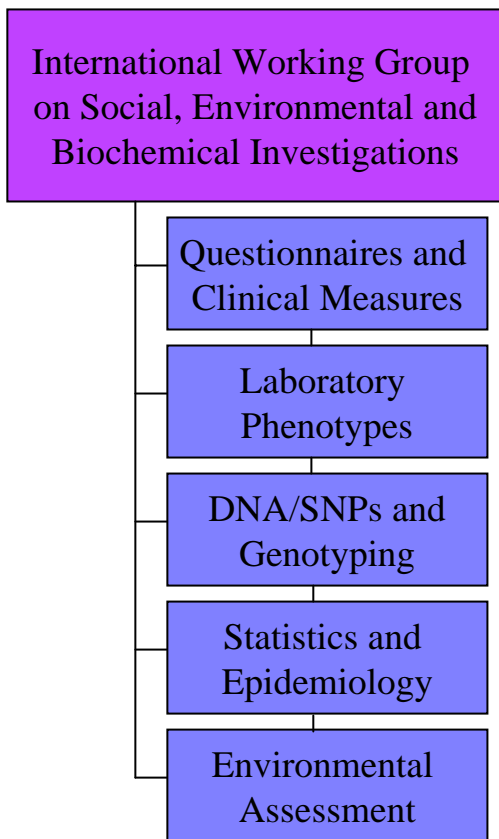
- ❖ International and Multidisciplinary Team
- ❖ Assures coordination among cores via quarterly discussions, annual meeting
- ❖ Sets priorities for cores

Cores

- ❖ Principal work units of P3G, focused on specific issues related to biobanks.
- ❖ In most (but not all) cases, cores are hosted at a single site and are integrated with an existing team actively involved with a biobank.



International Working Group on Social, Environmental and Biochemical Investigations



- ❖ Early Focus on Risk Factors for Cardiovascular Phenotypes in order to capitalize on existing expertise, yet face new opportunities made available by new technologies in phenotyping, environmental measures, genomics, etc.
- ❖ 6 month goals - collate information related to existing studies, available phenotype information, etc., and deposit in P3Gdb
- ❖ Interim Leader: Erich Wichmann (Germany)



Selected notes in regards to the Social/ Environmental/Biochemical Working Group

Questionnaires and
Clinical Measures

- ❖ Collation of information related to existing studies
- ❖ Evaluation of socio-demographic information, collected for public health purposes
- ❖ Definition of clinical phenotypes with different degree of complexity

Laboratory
Phenotypes

- ❖ Short term Focus: Storage of plasma, serum and urine for downstream studies
- ❖ Research Focus on intermediate phenotypes, including new methodologies in proteomics and metabolomics

DNA/SNPs and
Genotyping

- ❖ Quality assessment measures related to DNA and genotyping
- ❖ Tracking of risk alleles and haplotypes

Statistics and
Epidemiology

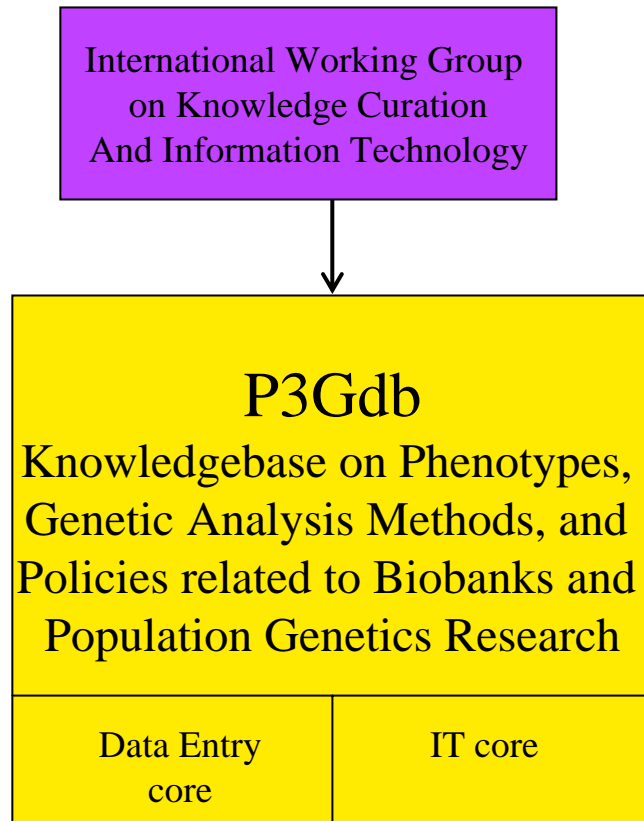
- ❖ Research into methods that allow merging and comparing population genetics studies that offer more power than “reducing” the data to a small set of “common” phenotypes

Environmental
Assessment

- ❖ Evaluation of environmental parameters needed for population genetics
- ❖ Definition of a core set of environmental information for studies of gene-environment interaction



International Working Group on Knowledge Curation And Information Technology



- ❖ P3G db is needed, as there is no existing resource that captures and disseminates information on large-scale epidemiological studies. Most of the details regarding questionnaires, phenotypes, methods are not available electronically (or at all), making comparative analyses difficult, and hindering meta-analyses (that could increase power, generate new hypotheses, etc.).
- ❖ “OMIM”-type resource.
- ❖ All activities of P3G, from all cores, must be organized and disseminated widely
- ❖ Multidisciplinary expertise required in informatics and communication.
- ❖ Data entry will be prioritized by working groups and cores.
- ❖ Immediate scientific benefits: Increased interactions between and beyond P3G as soon as P3Gdb is operational
- ❖ Interim Leader: Jan-Eric Litton (Sweden)



International Working Group on Ethics, Governance and Public Engagement

International Working Group
on Ethics, Governance and
Public Engagement

Population Genetics
And Policymaking

Impact on
Health Care Systems

Participation: Gender
Age, Ethnic Differences

- ❖ Public Engagement, Ethics, Public welfare (prevention/promotion) need to be integrated
- ❖ Interim Leader: Béatrice Godard (Canada)

- ❖ Policy issues not only related to the development of biobanks, but the translation of new data for the benefit of the public

- ❖ What risks and benefits will arise due to current practices in commercializing genetic information

- ❖ Research into practices used for public engagement
- ❖ Better understanding of public perception as they relate to socio-cultural issues



Challenges and Opportunities in Funding P3G

Challenges:

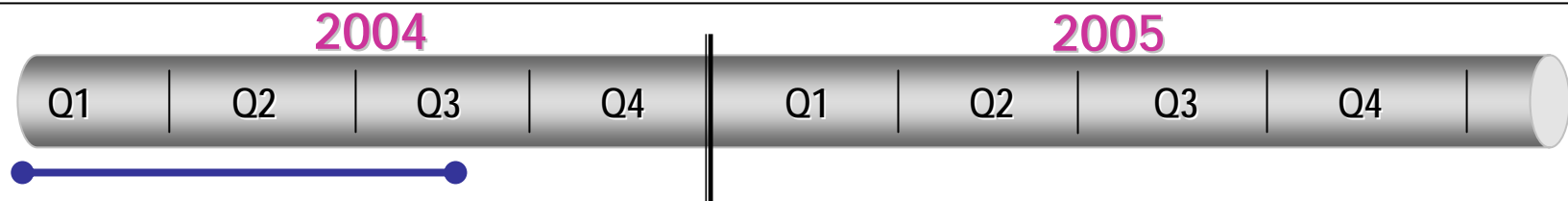
- ❖ Multi-national research initiatives are difficult due to limitations in funding research beyond national/continental boundaries.
- ❖ Biobanks are expensive - additional costs for international coordination are difficult to attract when biobank groups are working at raising funds to initiate or maintain a national Biobank.

Opportunities:

- ❖ Scientific benefits: Increased power afforded by large-scale collaboration, larger cohort-sizes for gene-environment studies, rarer phenotypes or subsets of participants with value-added phenotypes.
- ❖ Cost efficiencies: New knowledge (such as lower cost laboratory phenotyping methods) can decrease costs of biobank projects.
- ❖ Funding agencies may be more willing to fund portions of P3G activities (such as a core, for example) in the context of a larger effort with international co-funding).



Timeline (High Level)



Phase 1 – Planning and Funding

- Memorandum of Understanding completed
- Development of membership rules is complete
- Communication globally with participating projects and enrolment of new members are ongoing
- Writing of a detailed project plan for funding is in progress
- Securing of funding for global operations is ongoing
- Work plan for 2004 to 2006 has been drafted

Phase 2 – Implementation

- Creation of Board, Secretariat and Coordinating Committee
- Creation of detailed implementation plan for knowledge base
- Creation of forecast/deliverables tracking system
- Continue to enroll new members

Phase 3 – Deliverables

- Strengthen communications
- Continue to enroll new members
- Collection of common data
- Promotion, marketing, communications



Next Steps:

- Build Knowledge Database
- Membership drive
- Identify Remaining Cores
- Funding (Major) Applications
- Canadian Cohorts Meeting (May, 2005)
- IWG: Ethics, Governance, Public Engagement Workshop (Sept. 2005)
- Board of Directors Meeting (U.K., Sept. 2005)